



HIVEP2 gene

human immunodeficiency virus type I enhancer binding protein 2

Normal Function

The *HIVEP2* gene provides instructions for making a protein that functions as a transcription factor. Transcription factors attach (bind) to specific regions of DNA and help control the activity (expression) of particular genes. The HIVEP2 protein is most abundant in the brain, where it controls the expression of multiple genes, many of which are involved in brain growth and development. This protein may also play a role in the function of immune system cells and the process of bone remodeling, in which old bone is broken down and new bone is created to replace it. It may also be involved in other body processes; however these additional roles are not completely understood.

Health Conditions Related to Genetic Changes

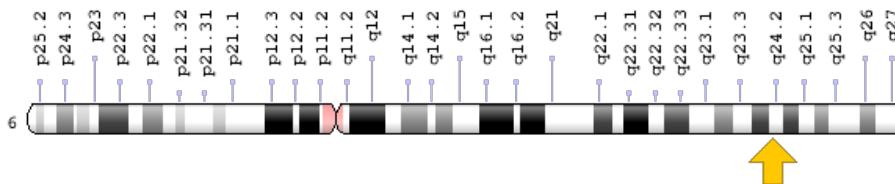
HIVEP2-related intellectual disability

At least nine mutations in the *HIVEP2* gene have been found in individuals with a neurological disorder called *HIVEP2*-related intellectual disability. This condition is characterized by delayed development of speech and walking, moderate to severe intellectual disability, mild physical abnormalities (dysmorphic features), and weak muscle tone (hypotonia), among other features. The *HIVEP2* gene mutations are thought to lead to a shortage of functional HIVEP2 protein. It is unclear how these genetic changes result in the features associated with *HIVEP2*-related intellectual disability, although researchers speculate that a shortage of the HIVEP2 protein alters the expression of several genes involved in brain growth and development. Abnormalities in the growth and development of the brain likely underlie the cognitive problems and other neurological features of *HIVEP2*-related intellectual disability. It is unclear how *HIVEP2* gene mutations contribute to the unusual physical features and health problems that can occur with this condition.

Chromosomal Location

Cytogenetic Location: 6q24.2, which is the long (q) arm of chromosome 6 at position 24.2

Molecular Location: base pairs 142,751,467 to 142,945,201 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- c-myc intron binding protein 1
- HIV-EP2
- MBP-2
- MHC binding protein-2
- MIBP1
- MRD43
- Schnurri-2
- SHN2
- ZAS2
- ZNF40B

Additional Information & Resources

Scientific Articles on PubMed

- PubMed

<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HIVEP2%5BTIAB%5D%29+OR+%28human+immunodeficiency+virus+type-I+enhancer+binding+protein+2%5BTIAB%5D%29%29+OR+%28%28HIV-EP2%5BTIAB%5D%29+OR+%28MBP-2%5BTIAB%5D%29+OR+%28MHC+binding+protein-2%5BTIAB%5D%29+OR+%28MIBP1%5BTIAB%5D%29+OR+%28SHN2%5BTIAB%5D%29+OR+%28Schnurri-2%5BTIAB%5D%29+OR+%28ZAS2%5BTIAB%5D%29+OR+%28c-myc+intron+binding+protein+1%5BTIAB%5D%29+OR+%28transcription+factor+HIVEP2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- HUMAN IMMUNODEFICIENCY VIRUS TYPE 1 ENHANCER-BINDING PROTEIN 2
<http://omim.org/entry/143054>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=HIVEP2%5Bgene%5D>
- HGNC Gene Family: Zinc fingers C2H2-type
<http://www.genenames.org/cgi-bin/genefamilies/set/28>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4921
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3097>
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